

Genetic testing can provide powerful medical insights. Selecting the right test is the first step.

DIAGNOSTIC GENETIC TESTING can help identify and manage an existing hereditary condition, provide insight into unexplained symptoms, and gauge the risk of developing a condition that runs in the family.

PROACTIVE GENETIC TESTING is an emerging area in clinical genetics that gives healthy adults the opportunity to gain insights about health-related risks based on their DNA. It is appropriate for healthy individuals without symptoms or a strong family history of disease.

The following flowchart can help determine whether your patient would benefit from diagnostic or proactive genetic testing with Invitae.

1 Take a personal and family medical history

A detailed personal and family medical history will help uncover information that may help guide your decision. Be sure to ask questions such as:

- Any major medical problems in the family?
- Any sudden or unexpected death in the family?
- Any early death in the family?

DIAGNOSTIC

PROACTIVE

2 Determine whether diagnostic or proactive testing is appropriate

If your patient has any of the following, diagnostic testing may be appropriate:

- A personal history of a condition that can have a genetic basis
- Strong family history of a condition that can have a genetic basis
- Symptomatic features or a suspected diagnosis for a condition that can have a genetic basis

If your patient is a healthy adult with any of the following, proactive genetic testing may be appropriate:

- No personal or family history of cancer or cardiovascular conditions
- Family history of cancer or cardiovascular conditions is limited or does not meet diagnostic criteria
- Uncertain or unknown family medical history

3 Test medically actionable genes

In diagnostic testing, Invitae's results indicate genetic variants that are pathogenic, likely pathogenic, or variants of uncertain significance. Report findings are stated in these diagnostic terms.

In proactive testing, Invitae's results can be negative (no found variants) or can identify pathogenic or likely pathogenic variants. Report findings are stated in predisposition terms (risk).

4 Develop appropriate next steps, based on test results

Receiving a diagnostic genetic diagnosis can have implications for treatment decisions, medical management options and outcomes, and can impact other family members.

Proactive testing can reveal an increased predisposition for certain conditions. Detecting these risks early can help guide preventive measures to stay healthy.

STILL UNCERTAIN?

Invitae's board-certified genetic counselors are available to help you order the right test and guide you through results interpretation and management. For more information or if you're ready to consult with one of Invitae's genetic counselors, please email clinconsult@invitae.com or call Client Services at 800-436-3037.